GENESYS Diagnostics INC.

Women's Health: Carrier Screening and Non-Invasive Prenatal Testing (NIPT) Requisition Form

*Information required for testing

Patient Information

LAST NAME*	FIRST NAME*	MI	DOB* (MM/DD/YYYY)	SEX

ADDRESS	CITY	STATE	ZIPCODE	PHONE NUMBER	EMAIL ADDRESS

Billing Information (Please include a copy of insurance card(s) for billing purposes.)

*☐ CLIENT BILL ☐ INSURANCE ☐ SELF PAY ☐ MEDICARE/MEDICAID (☐ PRIMARY ☐ SECONDARY) RELATIONSHIP: ☐ SELF ☐ SPOUSE ☐ DEPENDENT

INSURANCE NAME	MEMBER/POLICY ID	GROUP #

POLICY HOLDER NAME	POLICY HOLDER DOB (MM/DD/YYYY)

Account Information

FACILITY/PRACTICE NAME*	PHONE NUMBER	FAX NUMBER	ORDERING PHYSICIAN NAME*

Background Information (Please check all that apply)

RACE AND ETHNICITY: ☐ WHITE ☐ ASIAN ☐ HISPANIC ☐ AFRICAN AMERICAN ☐ ASHKENAZI JEWISH ☐ OTHER (PLEASE SPECIFY):_____

Carrier Screening

GENE PANEL NAME* (Must choose at least one)
☐ COMPREHENSIVE PANEL (145 GENES)
☐ HIGH-FREQUENCY PAN-ETHNIC PANEL (11 GENES)
☐ FRAGILE X (1 GENE)
☐ DUCHENNE MUSCULAR DYSTROPHY (1 GENE)
☐ SPINAL MUSCULAR ATROPHY (1 GENE)
☐ ALPHA THALASSEMIA (2 GENES)
☐ CYSTIC FIBROSIS (1 GENE)
☐ INDIVIDUAL GENE TESTING

SPECIMEN TYPE: ☐ BLOOD IN EDTA (5ml MIN) ☐ BUCCAL SWAB ☐ DNA (10 ug MIN)

Other ICD-10 Code_____

ICD 10 CODES*			
☐Z13.0	Encounter for screening for diseases of the blood and blood-forming organs and certain disorders involving immune mechanism	☐Z34.03	Encounter for supervision of normal first pregnancy, third trimester
☐Z13.228	Encounter for screening for other metabolic disorders	☐Z34.81	Encounter for supervision of other normal pregnancy, first trimester
☐Z13.71	Encounter for nonprocreative screening for genetic disease carrier status	☐Z34.82	Encounter for supervision of other normal pregnancy, second trimester
☐Z13.89	Encounter for screening for other disorder	☐Z34.83	Encounter for supervision of other normal pregnancy, third trimester
☐Z14.8	Other genetic carrier status	☐Z81.0	Family history of intellectual disabilities
☐Z15.89	High risk ethnicity	☐Z84.3	Consanguinity
☐Z31.430	Encounter of female for testing for genetic disease carrier status for procreative management	☐Z84.81	Family history of carrier of genetic disease
☐Z34.01	Encounter for supervision of normal first pregnancy, first trimester	☐Z84.89	Family history of other specified conditions
☐Z34.02	Encounter for supervision of normal first pregnancy, second trimester	☐Z84.99	Family history of related disorder. Please describe:_____

Non-Invasive Prenatal Testing (NIPT) 5ml Blood in Streck tube required

Must choose either single pregnancy or twin pregnancy*

☐ **SINGLE PREGNANCY: CHROMOSOMES 13, 18, 21 and sex chromosome aneuploidies (MX, XXX, XXY, and XYY)**

ADDITIONAL OPTION(S) SINGLE PREGNANCY ONLY (Insurance coverage requirements vary for additional options)

☐ **Microdeletions:** 1p36 deletion, 4p- (Wolf-Hirschhorn syndrome), 5p- (cri-du-chat syndrome), 15q11.2 (Prader-Willi syndrome/Angelman syndrome), 22q11.2 deletion (DiGeorge syndrome)

☐ **All chromosomes** (Trisomies of chromosomes 1-22 including sex chromosome aneuploidies [MX, XXX, XXY, and XYY])

☐ **TWIN PREGNANCY: CHROMOSOMES 13, 18, 21 and presence of Y chromosome**

GESTATIONAL AGE* (WEEKS, DAYS)	AS ESTIMATED ON (MM/DD/YYYY)	DATE OF DRAW* (MM/DD/YYYY)

DATING METHOD:* ☐ LMP ☐ DATE OF IMPLANTATION ☐ CRL ☐ OTHER

SPECIFY_____

ICD 10 CODES*
☐ Advanced maternal age (AMA), 1st pregnancy (O09.519, O09.511, O09.512, O09.513)
☐ Advanced maternal age (AMA), not 1st pregnancy (O09.529, O09.521, O09.522, O09.523)
☐ Abnormal ultrasound, non-CNS (O28.3)
☐ Abnormal ultrasound, CNS (O35.0XX0)
☐ Abnormal maternal screen (O28.3)
☐ Chromosomal abnormality suspected in fetus (O35.1XXO)
☐ Previous pregnancy/child affected with chromosome abnormality (O35.2XX0)
☐ Family history (Z84.89)
☐ Supervision, other high-risk pregnancy (O09.899, O09.891, O09.892, O09.893)
☐ Supervision, normal 1st pregnancy (Z34.00, Z43.01, Z34.02, Z34.03)
☐ Supervision, other normal pregnancy (Z34.80, Z34.81, Z34.82, Z34.83)
☐ Low risk/ Maternal anxiety
☐ Other_____

This test includes fetal sex. If you would like to omit fetal sex, you must check the box below.

☐**Omit fetal sex**

Patient Authorization (Please see patient informed consent on reverse side)

I understand that I am responsible for providing accurate information about my insurance to Genesys Diagnostics Inc. I understand that Genesys Diagnostics Inc. will be providing testing services and billing my insurance. However, I understand that charges that are not covered by my insurance, including any applicable co- payments and deductibles, are my responsibility and I agree to pay such charges promptly.

Patient/Guardian Signature:* Date:*

☐ I do not consent to having my deidentified DNA sample used for internal research purposes.

Healthcare Provider Authorization

I certify that (i) this test is medically necessary, (ii) the patient (or authorized representative on the patient's behalf) has given informed consent (which includes written informed consent or written authorization when required by law) to have this testing performed, and (iii) the informed consent obtained from the patient meets the requirements of applicable law. I agree to provide Genesys, or its designee, any and all additional information reasonably required for this testing to be performed.

Healthcare Provider Signature:* Date:*

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CARRIER SCREENING

COMPREHENSIVE PANEL (145 GENES): ABCA3, ABCC8, ABCD1, ACADM, ACADS, ACADVL, ACAT1, ACSF3, AFF2, AGA, AGXT, AHI1, AIRE, ALDOB, ALMS1, ALPL, ANO10, ARSA, ARX, ASL, ASPA, ATM, ATP7B, BBS1, BBS2, BCKDHA, BCKDHB, BLM, BTD, CAPN3, CBS, CC2D2A, CCDC88C, CDH23, CEP290, CFTR, CHRNE, CLCN1, CLRN1, CNGB3, COL7A1, CPT2, CYP11A1, CYP1B1, CYP21A2, CYP27A1, CYP27B1, DBT, DHCR7, DHDDS, DLD, DMD, DNAH5, DYNC2HI, DYSF, ELP1, ERCC2, EVC2, EYS, F11, F8, F9, FAH, FANCA, FANCC, FANCG, FKRP, FKTN, FMO3, FMR1, FXN, G6PC, GAA, GALC, GALT, GBA, GBE1, GJB2, GLA, GNE, GNPTAB, GRIP1, HBA1, HBA2, HBB, HEXA, HFE, HOGA1, HPS1, HPS3, IDUA, L1CAM, LDLR, LOXHD1, LRP2, MCCC2, MCOLN1, MCPH1, MEFV, MID1, MLC1, MMACHC, MMUT, MVK, MYO7A, NAGA, NEB, NPC1, NPC2, NPHS1, NPHS2, NR0B1, OCA2, OTC, PAH, PCDH15, PEX6, PKHD1, PLP1, PMM2, POLG, PRF1, PYGM, RARS2, RMRP, RNASEH2B, RPGR, RS1, SCO2, SERPINA1, SLC12A3, SLC19A3, SLC22A5, SLC26A2, SLC26A4, SLC37A4, SLC6A8, SMN1, SMPD1, TF, TMEM216, TNXB, TYR, USH2A, XPC FMR1

HIGH-FREQUENCY PAN-ETHNIC PANEL (11 GENES): HBA1, HBA2, HBB, PMM2, CFTR, DMD, FMR1, ACADM, PAH, DHCR7, SMN1

FRAGILE X (1 GENE): FMR1

DUCHENNE MUSCULAR DYSTROPHY (1 GENE): DMD

SPINAL MUSCULAR ATROPHY (1 GENE): SMN1

ALPHA THALASSEMIA (2 GENES): HBA1, HBA2

CYSTIC FIBROSIS (1 GENE): CFTR

Medical Necessity Statement: Tests ordered on Medicare patients must follow CMS rules regarding medical necessity and FDA approval guidelines and must include diagnosis, symptoms and reason for testing as indicated in the medical record. If testing does not come under Medicare guidelines for payment a 'signed' Advanced Beneficiary Notice must be included.

**Certain regions in various genes have poor coverage and are not included in the panel (if you would like more coverage information regarding any specific genes of interest, please contact Genesys Diagnostics Inc.). All genes that have pseudogenes will have poorer performance on the MiSeq instrument. Variants in genes with pseudogenes may not be reliably detected. DNA alterations in regions not covered by this test such as deep intronic or regulatory regions, or in poorly covered regions will not be detected using Next Generation Sequencing analysis. There are technical limitations on the ability of Next Generation Sequencing to detect small insertions and deletions and these types of alterations are not detected as reliably as single nucleotide variants. This assay is not designed or validated for the detection of low-level mosaicism or somatic mutations.

Patient Informed Consent Carrier Screening

It has been explained to me and I understand that I am voluntarily providing a specimen for a genetic test. I will provide the specimen in a collection device provided by GDI. My DNA will be extracted from my specimen at GDI, and the test will evaluate how my genome variance may affect the risk associated with genetically linked disorders. However, carriers for certain disease genes or variants may show mild phenotypes themselves. Proper pre-test and post-test genetic counseling should be provided. The test identifies the most common variants of these genes but is not designed to identify some rare mutations. The test is a clinical laboratory test and may aid in my treatment plan; therefore, I or my health insurer will be billed for this test. A written report of the test results will be provided to my health care provider who will inform me of the results. GDI will keep all of my medical information confidential and only disclose it to pursuant to applicable state and federal laws. I understand that I am responsible for providing accurate information about my insurance to Genesys Diagnostics Inc. I understand that Genesys Diagnostics Inc. will be providing testing service and billing my insurance. However, I understand that charges that are not covered by my insurance, including any applicable copayments and deductibles are my responsibility and I agree to pay such charges promptly.

Patient Informed Consent NIPT

INTRODUCTION: This form describes the benefits, risks, and limitations of this screening test. You should seek pre-test counseling by a genetic counselor or other experienced health care provider prior to undergoing this test. Read this form carefully – and ask any questions you may have of your health care provider -- before making your decision about testing.

PURPOSE: The purpose of this test is to screen your pregnancy for certain chromosomal abnormalities, also known as "aneuploidies." This test gives information about whether there may be extra copies (trisomy) of chromosomes 21, 18, and 13, and the option to know if there is an extra copy of a sex chromosome (X or Y), and/or a missing copy of sex chromosome (thMX). Fetal sex may also be reported. This test has the option to screen for aneuploidies (extra copies) in all chromosomes. In addition, the option to screen for the following microdeletion (small, missing parts of chromosomes) syndromes: 1p36 deletion, 4p- (Wolf-Hirschhorn syndrome), 5p- (cri-du-chat syndrome), 15q11.2 (Prader-Willi syndrome/Angelman syndrome), 22q11.2 deletion (DiGeorge syndrome or velocardiofacial syndrome) is also available. For chromosomes 21, 18, and 13, the this test is validated in singleton and twin pregnancies. In twin pregnancies, sex chromosome testing can only screen for the presence or absence of the Y chromosome, and not for extra or missing sex chromosomes. This test can be performed as early as 10 weeks 0 days gestational age. Consult your health care provider if you would like more information about this screening test, including risks, limitations, performance data, error rates, descriptions of the conditions being screened, and what these results may mean to your pregnancy.

HOW THIS TEST WORKS: This test screens for specific chromosomal abnormalities by looking at the DNA (genetic material) in your blood. The sample of blood includes a combination of both your DNA and the DNA from the pregnancy. A technology called massively parallel sequencing is used to count the amount of DNA from each test chromosome and/or from specific regions of chromosomes. The laboratory then uses an analysis method to determine if each of the conditions you have elected to test for is likely to be present or absent.

SEX OF PREGNANCY: Depending upon the option you and your health care provider elect, the test results may include the sex of the pregnancy. If you do not wish to know the sex, please tell your health care provider not to disclose this information to you. Depending upon the test ordered, you may not be able to prevent learning the sex of your pregnancy. In rare instances, incorrect sex results can occur.

LIMITATIONS OF THE TEST: These are screening tests that look only for specific chromosomal abnormalities. This means that other chromosomal abnormalities may be present and could affect your pregnancy. A "No Aneuploidy Detected" result does not guarantee a healthy pregnancy or baby and does not eliminate the possibility that your pregnancy may have birth defects, genetic conditions, or other conditions, such as open neural tube defects or autism.

There is a small possibility that the test results might not reflect the chromosomes of the fetus, but may reflect chromosomal changes of the placenta (confined placental mosaicism, CPM) or of you (maternal chromosomal abnormalities). While these tests are not designed to assess your health, in some cases, information about your health may be revealed directly or indirectly (e.g., when combined with other information). Examples include maternal XXX, sex chromosome status or benign or malignant maternal neoplasms. In a twin pregnancy, the status of each individual fetus cannot be determined.

These tests, like many tests, have limitations, including false negative and false positive results. This means that the chromosomal abnormality being tested for may be present even if you receive a negative result (this is called a 'false negative'), or that you may receive a positive result for the chromosomal abnormality being tested for, even though the abnormality is not actually present (this is called a 'false positive').

In the case of a twin pregnancy, the presence or absence of Y chromosome material can be reported. The occurrence of sex chromosome aneuploidies cannot be evaluated in twin pregnancies. In the case of a vanishing twin, the test result may reflect the DNA of the vanishing twin, leading to a higher probability of false positive or false negative results.

No irreversible clinical decisions should be made based on these screening results alone. If definitive diagnosis is desired, chorionic villus sampling or amniocentesis would be necessary. In some cases, other testing may also be necessary. Some rare chromosomal aneuploidies may only occur in mosaic form. Clinical consequences depend on the chromosome involved and can not be predicted prenatally.

Consult your health care provider for more information about your results and what they may mean for your pregnancy, what options you will have for further testing, and whether additional testing is recommended for you based on your clinical history.

TEST PROCEDURE: A tube of your blood will be drawn and analyzed.

PHYSICAL RISKS: Side effects of having blood drawn are uncommon, but may include dizziness, fainting, soreness, bleeding, bruising, and, rarely, infection.

DISCRIMINATION RISKS: Genetic information could be used as a basis of discrimination. To address concerns regarding possible health insurance and employment discrimination, some countries, U.S. states, and the U.S. government have enacted laws to prohibit genetic discrimination in those circumstances. The laws may not protect against genetic discrimination in other circumstances, such as when applying for life insurance or long-term disability insurance. Talk to your health care provider or genetic counselor if you have concerns about genetic discrimination prior to testing.

PREGNANCY OUTCOME INFORMATION: Collecting information on your pregnancy after testing is part of a laboratory's standard practice for quality purposes and is required in several states. As such, Genesys or its designee may contact your health care provider to obtain this information. By executing this informed consent, you agree to allow your health care provider to provide this information to Genesys or its designee.

SECONDARY FINDINGS: In the course of performing the analysis for the indicated tests, information regarding other chromosomal alterations, also known as "secondary findings" may become evident. Our policy is to NOT REPORT any secondary findings that may be noted in the course of analyzing the test data.

PRIVACY: Test results are kept confidential. Your test results will only be released in connection with the testing service, to your health care provider, his or her designee, other health care providers involved in your medical care, or to another health care provider as directed by you (or a person legally authorized to act on your behalf) in writing, or otherwise as required or authorized by applicable law.

USE OF INFORMATION AND LEFTOVER SPECIMENS: Pursuant to best practices and clinical laboratory standards, leftover de-identified specimens (unless prohibited by law), as well as de-identified genetic and other information learned from your testing, may be used by Genesys or others on its behalf for purposes of quality control, laboratory operations, laboratory test development, and laboratory improvement. All such uses will be in compliance with applicable laws. Leftover specimens from New York State will be destroyed within 60 days.

RESEARCH: We may use your leftover specimen and your health information, including genetic information, in a de-identified form (unless otherwise allowed by applicable law) for research purposes. Such uses may result in the development of commercial products and services. You will not receive notice of any specific uses and you will not receive any compensation for these uses. All such uses will be in compliance with applicable law. This does not apply to leftover specimens collected from New York State.

TEST RESULTS: Your test results will be sent to the health care provider. Your healthcare provider is responsible for interpreting the test results and explaining the meaning to you. Genesys does provide genetic counseling services directly to patients upon request.